Del (18p) syndrome with increased nuchal translucency revealed in prenatal diagnosis

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LETTER TO THE EDITOR

DEL (18p) SYNDROME WITH INCREASED NUCHAL TRANSLUCENCY REVEALED IN PRENATAL DIAGNOSIS

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Fetal chromosomal abnormality and increased nuchal translucency (INT) association is well established (6). There are several reports about the association between foetal chromosomal abnormalities including triploidy, trisomies 13, 18, 22, tetrasomy 12p and increased nuchal translucency (INT) during the first trimester of pregnancy (6). More than 70% of cases with trisomy 21 can be detected by screening for INT (8). In the present foetus with monosomy 18p, we observed INT as the only abnormal ultrasonographic finding during pregnancy.

Amniocentesis was performed at 18 weeks of gestation on a 41 years old Turkish female referred for prenatal diagnosis because of advanced maternal age. INT was observed at the time of amniocentesis (2.8 cm). The couple had a healthy four year-old girl with 46,XX karyotype. Conventional cytogenetic analysis of cultured amniocytes showed an unbalanced whole arm translocation between the long arm of one chromosome 18 and the long arm of one chromosome 22, 45,XX, der(18)t(18;22)(q10;q10), which led to monosomy 18p in the foetus (Fig. 1a). Parental karyotypes were normal, thus the aberration had occurred *de novo*. The derivative chromosome had one centromere revealed by conventional C banding staining. Whether the centromere originated from chromosome 18 or chromosome 22 could not be determined by FISH. Chromosome 18p STS markers D18S498, D18S481 and D18S170 were used for genotyping of the foetus and parents, to determine the origin of derivative chromosome 18. Marker D18S170 was informative: while her parents were heterozygous for this marker, the foetus had only the paternal allele, thus the maternal chromosome 18 was found to be involved in the translocation (Fig. 1b). The parents decided to terminate the pregnancy at 21 weeks of gestation based on the chromosomal abnormality. At autopsy, 362.62 g weight of foetus, supraorbital flatness, depressed nasal bridge, flared nostrils and low set ears were observed. Placenta weight was lower (170 g) than average

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for comparison with the autopsy findings given above (5). Our case has displayed no other clinical features except INT at the first trimester ultrasonographic examination. Strikingly in another previous study, a case with t(15;18) resulting in del(18p) also had only ultrasonographic INT finding (4).
Thus, our study supports that INT was helpful to perform prenatal cytogenetic analysis and also it illustrates the association between INT and chromosomal abnormalities, 18p monosomy.

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